

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 2. PAX2 Mutations Identified in Renal Coloboma Syndrome

Exon	Mutation original description	Mutation Current numbering from ATG	Protein Domain	Reference
2	insG619	c.70_79insG	Paired	Sanyanusin et al 1996, Schimmenti et al 1997, Schimmenti et al 1999, Amiel et al 2000, Porteous et al 2000, Ford et al 2001
2	insGG619	c.70_79insGG	Paired	Amiel et al 2000
2	delG619	c.70_79delG	Paired	Schimmenti et al 1999
2	delt602	c.59deIT	Paired	Chung et al 2001
2	del674-695	c.131_152del	Paired	Schimmenti et al 1997
2	delt611	c.68deIT	Paired	Cunliffe et al 1998
2	del682-691	c.139_148del	Paired	Fletcher et al 2005
2	G755C	c.212G>C	Paired	Higashide et al 2005
3	G719A	c.176G>A	Paired	Devriendt et al 1998
3	delG832	c.289delG	Paired	Amiel et al 2000
3	C886T	c.343C>T	Paired	Schimmenti et al 2003
3	del658-663	c.115_120del CAGCGC	Paired	Salomon et al 2001
3	763insgagac c	c.219_222dup GAGACC	Paired	Devriendt et al 1998
5	delC1104	c.561delC	Octapeptide	Sanyanusin et al 1995
7	C1297T	c.754C>T	Homeodomain	Porteous et al 2000
7	C1318T	c.775C>T	Homeodomain	Nishimoto et al 2001
9	C1566A 46,XY, t(10;13) (q24.3;q12.)	c.1023C>A	PST domain translocation between exons 3 and 4	Nishimoto et al 2001 Narahara et al 1997

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